Sarah B Pierce

List of Publications by Year
in descending order

10	823	1040056 9	1372567 10
papers	citations	h-index	g-index

10	10	10	1471
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
2	Helicase-inactivating BRIP1 mutation yields Fanconi anemia with microcephaly and other congenital abnormalities. Journal of Physical Education and Sports Management, 2020, 6, a005652.	1.2	2
3	De novo mutation in RING1 with epigenetic effects on neurodevelopment. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1558-1563.	7.1	24
4	Mutation of KREMEN1, a modulator of Wnt signaling, is responsible for ectodermal dysplasia including oligodontia in Palestinian families. European Journal of Human Genetics, 2016, 24, 1430-1435.	2.8	20
5	Infantile onset spinocerebellar ataxia caused by compound heterozygosity for Twinkle mutations and modeling of Twinkle mutations causing recessive disease. Journal of Physical Education and Sports Management, 2016, 2, a001107.	1.2	13
6	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. Neurology, 2014, 83, 2054-2061.	1.1	86
7	Mutations in LARS2, Encoding Mitochondrial Leucyl-tRNA Synthetase, Lead to Premature Ovarian Failure and Hearing Loss in Perrault Syndrome. American Journal of Human Genetics, 2013, 92, 614-620.	6.2	176
8	Mutations in mitochondrial histidyl tRNA synthetase HARS2 cause ovarian dysgenesis and sensorineural hearing loss of Perrault syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6543-6548.	7.1	225
9	Garrod's fourth inborn error of metabolism solved by the identification of mutations causing pentosuria. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 18313-18317.	7.1	11
10	Mutations in the DBP-Deficiency Protein HSD17B4 Cause Ovarian Dysgenesis, Hearing Loss, and Ataxia of Perrault Syndrome. American Journal of Human Genetics, 2010, 87, 282-288.	6.2	231